



NOTES

CARBOHYDRATE METABOLISM DISORDERS

GENERALLY, WHAT ARE THEY?

PATHOLOGY & CAUSES

- Inherited disorders of carbohydrate metabolism (fructose, glucose, galactose)
- Determined by exposure to external triggers (e.g. dietary factors)
- Disruption in certain metabolic pathways → accumulation of byproducts → systemic alterations, organ failures

SIGNS & SYMPTOMS

- See individual disorders

DIAGNOSIS

LAB RESULTS

- Enzymatic dosage

OTHER DIAGNOSTICS

- Genetic testing

TREATMENT

OTHER INTERVENTIONS

- Avoid triggers

ESSENTIAL FRUCTOSURIA

osms.it/essential-fructosuria

PATHOLOGY & CAUSES

- **Benign** disease; impaired fructose metabolism → increased excretion in urine

CAUSES

- **Autosomal recessive mutation in fructokinase**, AKA ketohexokinase (KHK)
 - First enzyme in fructose metabolism
- Intake of fructose-rich food/fructose-related sugar (e.g. sucrose, sorbitol) → increase of fructose in blood → excretion of **fructose in urine**

SIGNS & SYMPTOMS

- **Asymptomatic**

DIAGNOSIS

LAB RESULTS

- Positivity for reducing substances in urine

TREATMENT

OTHER INTERVENTIONS

- No treatment necessary

VISIT...

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GALACTOSEMIA

osms.it/galactosemia

PATHOLOGY & CAUSES

- Disease characterized by altered metabolism of galactose

CAUSES

- Autosomal recessive disorder

Three possible mutations

- Galactose-1-phosphate uridyl transferase (GALT) deficiency (most common)
 - Accumulation of galactose-1-phosphate + alternative metabolic pathway activation → accumulation of galactose metabolites (galactitol, galactonate)
- Galactokinase deficiency (uncommon)
 - Less severe, often asymptomatic
- Uridine diphosphate (UDP) galactose-4-epimerase deficiency (uncommon)

COMPLICATIONS

- Metabolites accumulate in
 - Eyes: increasing osmotic pressure → water attraction → opacification of lens (cataract)
 - Liver: fatty alteration, cirrhosis
 - Brain: cell damage, edema, gliosis
 - Kidney: prevent amino acid reabsorption → aminoaciduria
- Altered neutrophil activity → increased risk of sepsis from E.coli
- Gonadal failure, hemolysis, coagulopathy

SIGNS & SYMPTOMS

- FTT
- Vomiting, diarrhea with lactation
- Jaundice, hepatomegaly
- Cataract
- Intellectual disability, lethargy, speech disorder, altered muscle coordination (ataxia)

DIAGNOSIS

LAB RESULTS

- Routinely screened in newborns
 - Dosage of enzymes involved in galactose metabolism

TREATMENT

OTHER INTERVENTIONS

- Elimination of galactose from diet

HEREDITARY FRUCTOSE INTOLERANCE

osms.it/hereditary-fructose-intolerance

PATHOLOGY & CAUSES

- Disease characterized by alteration of fructose metabolism

CAUSES

- Autosomal recessive mutation in liver aldolase (aldolase B)
- Aldolase B deficiency → fructose 1-phosphate buildup →
 - Local fructose toxicity → liver cell death
 - Hypoglycemia
 - Adenosine triphosphate (ATP), phosphate depletion → inhibition of protein synthesis → hepatic, renal damage

SIGNS & SYMPTOMS

- Avoidance of foods containing fructose (e.g. fruit, juice)
- Acute ingestion of fructose/compounds which contain fructose (e.g. sucrose, sorbitol) → nausea, vomiting; restlessness; pallor; sweating, trembling; lethargy, apathy; convulsions; coma
- Repeated ingestion of fructose → failure to thrive (FTT); liver disease (e.g. jaundice, hepatomegaly); kidney disease

DIAGNOSIS

OTHER DIAGNOSTICS

- Nutritional history, clinical presentation
- Genetic testing

TREATMENT

OTHER INTERVENTIONS

- Elimination of fructose from diet